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The "Affected" Post-Preimplantation Genetic Diagnosis Embryo¹

Estair Van Wagner, Roxanne Mykitiuk & Jeff Nisker

Introduction

The meaning of "health" is constructed from a variety of perspectives, including biomedical, social and political, and in a variety of sites, including human bodies and natural environments. In this chapter we suggest that the human embryo is one such site. At first glance the *in vitro* embryo is not an obvious location from which to examine such constructions; however, we contend that an increasing focus on biomedical determinations of the "health" of the human embryo (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008) is significant not only in the application to human embryos themselves, but also in terms of our broader understanding of "health" in relation to existing adults and children.

New technologies and research initiatives are shaping the way in which we look at the embryo and what we look for (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008). Conventionally, the term "embryo" denotes the product of fertilisation of a human oocyte by a human sperm generally until eight weeks' development (Warnock, 1984). Numerous groups and individuals have attempted to characterise and describe the human embryo from perspectives such as

1 The authors are grateful to the CIHR Institute of Child and Youth Health and Human Development for funding this research.

ethics, religion, science or medicine, in relation to the objectives and interests of their respective communities (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008). In Australia, the legal definition² of "human embryo" is "a compromise between different views and resulted from the legal imperative to have a defined point against which legal judgments could be made" (Australian Government, 2005: 173). Heightened interest in defining and characterising the human embryo has resulted from the creation and manipulation of embryos outside of women's bodies, particularly through research related to assisted reproduction (Blake, Proctor, Johnson et al, 2002; Steptoe and Edwards, 1978; Yuzpe, Brown, Casper et al, 1989) and genetic testing (Handyside, Kontogianni, Hardy et al, 1990; Verlinsky, Lifchez, Valle et al, 1990; Verlinsky, Handyside, Simpson et al, 1993; Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008).

The characterisation of human embryos affects the ways embryos may be used, by women undergoing *in vitro* fertilisation (IVF), or by clinicians and scientists (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008). For instance, researchers and clinicians have emphasised selection of the "best" or "most suitable" embryo for implantation to achieve the highest pregnancy rate while removing the risk of high order multiple pregnancy (Mykitiuk and Nisker, 2008a; Min, Claman and Hughes, 2006). As assisted reproductive technology (ART) increasingly employs genetics-based techniques such as pre-implantation genetic diagnosis (PGD), new characterisations of the human embryo will emerge based on the new information made available (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008). PGD facilitates the selection of embryos created by IVF for transfer to the woman based on the particular criteria being tested such as the presence of genetic

2 This statement was made in relation to the definition of "human embryo" in the *Research Involving Human Embryos Act 2002* (Cth), which has since been amended by the *Prohibition of Human Cloning for Reproduction and the Regulation of Human Embryo Research Amendment Act 2006* (Cth), Sch 2 [2]. The point about the dependency of the legal definition of the human embryo on different contexts remains relevant, and perhaps is even reinforced by the context in which these changes were made (see the discussion in Australia's *Lockhart Review* (Australian Senate Committee on Community Affairs, 2006)).

markers for diseases (Handyside, Kontogianni, Hardy et al, 1990; Nisker and Gore-Langton, 1995) or a compatible tissue match for an existing sick sibling (Verlinsky, Rechitsky, Schoolcraft et al, 2001). Further, as new research initiatives such as stem cell research develop, policy-makers may characterise human embryos in such a manner as to promote research (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008). The purpose of this chapter is to examine the language employed in recent policy statements and regulatory documents to characterise the "affected" post-PGD embryo and to discuss the interdependent relationship between how embryos are characterised and the uses and purposes which PGD serves. In this examination we illustrate how understandings of "health" are produced through the research uses and clinical practice of PGD (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008).

Background, Methods and Characterisations

Before embryo cryopreservation (Trounson and Mohr, 1983), determinations of embryo "health" were based on morphologic criteria, in other words, by looking at the embryo through a microscope. Clinicians would look for features such as cell division, absence of fragmentation, and blastomere symmetry and clarity. These observations led to the selection of the "best" three or more embryos, which were transferred into the woman, the remaining embryos being discarded (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008). Within the past 15 years, embryos not selected for transfer have been cryopreserved for later transfer so that the woman could avoid the risks associated with menotropin drugs (Abramov, Elchalal and Schenker, 1999) and oocyte retrieval surgeries (Alsalili, Yuzpe, Tummon et al, 1995), either of which may be employed in additional IVF cycles (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008). At some IVF clinics today, clinicians and scientists still use microscopic criteria to determine which embryos are the "healthiest-looking", and transfer the "best" embryos while "fresh", in order to achieve the highest pregnancy rate (Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008). In fact there is no evidence that

an embryo's potential to become a child can be conclusively determined using morphologic characteristics viewable through a microscope. There is in fact evidence to the contrary (Tekpetey, Hughes, Shepherd et al, 2003).

The number of gene markers identified through PGD is rapidly expanding and provides another context in which determinations of the "best" or "healthy" embryo are being made (Lau and Leung, 2005). Such biomedical characteristics may be used to prevent the birth of a child who may suffer from a particular perceived "health" problem, or to positively select characteristics of a potential child (Levy, 2002; Mykitiuk and Nisker, 2008b; Van Wagner, Mykitiuk and Nisker, 2008). The focus of this chapter is on the "post-PGD embryo" – those embryos that have been tested using PGD. As our analysis demonstrates, the genetic information available through PGD and the intention behind diagnosis lead to several possible categorisations of the post-PGD embryo (Van Wagner, Mykitiuk and Nisker, 2008).

Our research focuses on four jurisdictions: the United Kingdom, Australia, Canada and New Zealand. We consider legislative, policy, scientific and research documents relating to PGD. These documents construct the regulation of PGD as part of the governance of ART – the use of *in vitro* human embryos. In the jurisdictions examined here, PGD is governed by "facilitative" legislative regimes, which establish "broad legislative frameworks" (Human Genome Research Project (HGRP), 2006: 302) in which decision-making powers are delegated to statutory and/or professional bodies. We argue that the documents examined here serve an important, but under-examined function in shaping the clinical practice and scientific application of PGD. Our examination will analyse the precise language through which characterisations of the post-PGD embryo occur. Further, we explore how resulting use or non-use of post-PGD embryos relate to understandings of "health" in the context of ART and embryo research (Van Wagner, Mykitiuk and Nisker, 2008).

Five possible characterisations of the post-PGD embryo emerge from our analysis. These characterisations include: 1) affected; 2) unaffected; 3) carrier; 4) sex-selected; and 5) HLA tissue-typed (Van Wagner, Mykitiuk and Nisker, 2008). The focus of this chapter is on the affected post-PGD embryo and how this determination is made in relation to what is considered

an unaffected post-PGD embryo.³ These characterisations are not mutually exclusive as the post-PGD embryo may exist within multiple categories at once, or change from one to another depending on the uses to which it may or may not be put.

The Unaffected Embryo

PGD enables clinicians to identify embryos as "affected" or "unaffected", allowing women undergoing IVF treatment to choose "embryos that are predicted to be unaffected" (HGC, 2006: 44) for implantation. This selection "provides an opportunity to begin a pregnancy knowing that only unaffected embryos have been transferred" (HFEA and ACGT, 2000: 8). The unaffected embryo is said to be "suitable" to be transferred or implanted in light of its status as "disease-free" (CBS, 2005a: 2), "without" (HGC, 2004: 10), or "free" (NECAHR, 2004: 3) of a genetic disorder or "chromosomal abnormality" (HGC, 2004: 18), not having "a copy of the faulty gene" (HFEA, 2005: 7), "not carry[ing] markers for the condition in question" (HCAHO, 2005: 1), "not known to have such an abnormality" (*HFE Bill*: s 14(4)(9)) or "not hav[ing] the particular gene mutation" (CBS, 2005a: 4).

After PGD is completed there may be more embryos than are required for immediate transfer. Current practice in some jurisdictions (ACART, 2006: 12; HGRP, 2006: 47, 52) is to keep the number of embryos transferred to a minimum. As PGD necessarily involves the creation of embryos for genetic analysis (CBS, 2005a: 4), one of its more controversial aspects is what happens to the embryos created which are not transferred to the woman. As with all IVF embryos, post-PGD "remaining" (CBS, 2005a: 5), "supernumerary" (CBS, 2005b: 2), "spare" (STC, 2005: 23; HGRP, 2006: 47), "surplus" (ACART, 2006: 13; HGRP, 2006: 47) or "excess" (*Research Involving Human Embryos Act 2002* (Cth) (RIHE Act) s 7(1)) embryos are those "no longer required" (Australian Government, 2005: 7) for the reproductive purposes for which they were created. The manner in which embryos are classified as one of the five characterisations differs across jurisdictions, as do the ways in which they may be used for

3 The authors explore all five themes in more detail elsewhere – see Van Wagner, Mykitiuk and Nisker, 2008.

reproductive or research purposes.⁴ Unaffected embryos "remaining" (CBS, 2005a: 33), could be "destroyed" (CBS, 2005a, 2005b; HGRP, 2006: 163), stored (STC, 2005: 23; HFEA, 2005: 7; *HFE Bill*: s 15(2)(b)(ab)),⁵ "used for research purposes" (HGRP, 2006: 163) or donated to "another individual" (STC, 2005: 23) for "reproductive purposes" (CBS, 2005b: 33; Van Wagner, Mykitiuk and Nisker, 2008).

While policy documents and legislation do not use "healthy" as a general descriptor of the unaffected embryo, they have used the term in discussions about the disposition of embryos not to be used for reproductive purposes, either because there are additional unaffected embryos, which are not needed for implantation, or because PGD is being employed to select a tissue-match or avoid a carrier embryo (HGC, 2006: 14, 51, 52; HGRP, 2006: 21, 43; NHMRC, 2007b: 42). These situations are framed as creating an ethical dilemma with respect to the disposal or use of "healthy", or unaffected embryos, which implies that the disposal or use of an affected post-PGD embryo does not pose the same ethical issues (Van Wagner, Mykitiuk and Nisker, 2008).

We contend that an assessment or characterisation of "health" based on the outcome of PGD is problematic because only specific, limited genetic markers are identified, and therefore, "health" is understood as the absence of these markers and the medical conditions they are associated with in living persons (Van Wagner, Mykitiuk and Nisker, 2008). "PGD is not a guarantee of a healthy baby" (HGRP, 2006: 17, 52), and as the Human Fertilisation and Embryology Authority (HFEA) in the UK has indicated, through the use of PGD, "the woman makes a decision about suitability based on information about the genetic status of the embryo" (2005: 9), which is only one of many factors in the overall health of a child.

The Affected Embryo

An embryo will be considered "affected" if the presence of a genetic "anomaly" (CBS, 2005a: 1), "mutation" (HGRP, 2006: 1)

4 For a more detailed account see Van Wagner, Mykitiuk and Nisker, 2008.

5 This section pertains to the storage of embryos generally.

or an "abnormality" (Australian Government, 2005: 120; *HFE Bill*: s 14(4)(9), Sch 2.5.3 para 1ZA) has been detected through PGD. Under the current practice of PGD, such information is used to identify embryos affected by "serious, life threatening conditions" (HGRP, 2006: 5), "a genetic disease" (ACART, 2006: 13), a "serious genetic disorder" (HGC, 2004), "serious genetic defects" (Australian Government, 2005), "genetic abnormality or disease" (ITA, 2006b), "serious genetic abnormality or a disease" (*Human Reproductive Technology Act 1991* (WA) s 14(2b)(a)(ii)) or "genetic conditions incompatible with life, or with a life of quality" (STC, 2005: 28). In the jurisdictions we examined, PGD is currently applied to diagnose X-linked conditions, "numerical chromosomal abnormalities" (CBS, 2005a: 4; HGRP, 2006: 3, 37; NECAHR, 2004: 4), specific gene mutations or "single-gene defects" (NECAHR, 2004: 4).

Defining "serious"

The term "serious" is invoked in a number of jurisdictions as a threshold between current and acceptable uses of PGD and those characterised as "trivial or [for] social reasons" (HGC, 2004: 19; HGRP, 2006: 46; Van Wagner, Mykitiuk and Nisker, 2008). Despite widespread use of the term in this way, there is no common or specific definition of "serious" in any of the four jurisdictions we examined (HCARO, 2005; HGC, 2004: 22; HGRP, 2006: 236; NECAHR, 2004: 9; NHMRC, 2007b: 42).

The HFEA's *Code of Practice* states that PGD will be offered "only where there is a significant risk of a serious genetic condition being present in the embryo" (HFEA, 2003: 123). The HFE Bill, being reviewed by Committee as of May 2008 would amend the *Human Fertilisation and Embryology Act 1990* (UK) to restrict "embryo testing" for "gene, chromosome or mitochondrial abnormality" and sex-linked conditions (*HFE Bill*: s 14(4)(9), s 14(4)(10)) to cases where "there is a significant risk that a person with the abnormality will have or develop a serious physical or mental disability, a serious illness or any other serious medical condition" (*Bill* Sch 2.5.3 para 1ZA(1)(a), (2)). In a 2005 report on the application of PGD for "lower penetrance susceptibility conditions", the HFEA stated, "[h]ow serious a condition is depends on how having the condition

affects, threatens or limits the life of the individual" (2005: 11). According to the report, a condition that will not "cause someone to suffer or detrimentally affect their life" would be "unlikely to be regarded as serious", while a condition that requires "regular invasive treatment, or was life-limiting or life threatening" would be (HFEA, 2005: 11). The HFEA has since announced a policy approving the use of PGD to detect the BRCA 1, BRCA 2, and HNPCC genes.⁶ This change arguably opened up a wider application of PGD than that allowed by the previous HFEA position, which had largely limited the practice to detection of high penetrance⁷ and early onset conditions (Krahn, 2007: 1445; Van Wagner, Mykitiuk and Nisker, 2008).

The HFEA does not provide a definition for "serious", instead leaving it to "discussion between the people seeking treatment and the clinical team" (HFEA, 2003: 123). It provides no formal list of "serious" conditions for which PGD is permitted, but in practice, because of its licensing procedure, particular PGD applications form an "accepted list of conditions" (STC, 2005). The *Code of Practice* outlines factors to be considered in determining when PGD is appropriate, including the perspective of the woman, or couple, the family situation, as well as the nature of the specific condition in question (HFEA, 2003: 123). While the original Draft Bill would have amended the HFEA Act to require the consideration of five factors in determining whether embryo testing is "necessary or desirable", neither the woman's nor her partner's perspective, nor their family circumstances were included (2007: s 59 Sch 2 para 1ZA(3)). This provision was not included in the HFE Bill now under consideration by Parliament.

In Australia the regulation of PGD falls under State jurisdiction; however, several States employ the Commonwealth regulatory regime on embryo research governed by the NHMRC's

6 See Human Fertilisation and Embryology Authority, "HFEA - Authority decision on PGD policy", available at <www.hfea.gov.uk/cps/rde/xchg/SID-3F57D79B-FDBD411B/hfea/hs.xsl/1124.html> (last accessed 28 May 2007).

7 The penetrance of BRCA mutations in the Ashkenazi Jewish community is very high. It is almost 80 per cent (Struewing, Abeliovich, Peretz et al, 1995; Tonin, Mes-Masson, Futreal et al, 1995; Warner, Foulkes, Goodwin et al, 1999).

Ethical Guidelines. This regime restricts PGD to conditions that "seriously harm" (NHMRC, 2007b: para 12.2). Some States do have specific legislation pertaining to ART and PGD. Western Australia's Reproductive Technology Council (RTC) states that the "seriousness of a genetic disease should be considered in the broad context of the environmental and personal factors of the participants" (RTC, 2004: 3). Licence applications for PGD contain the report of a "clinical geneticist" that addresses a number of factors including: the family's "experience with, and attitude to" the condition; the "level of impairment to body functions and structures that is usually associated"; the difficulties expected in "participating in activities such as learning and applying knowledge, communication, mobility, self-care, employment, community, social and civic life"; the "level of support" required and the "capacity of the family" to provide it; and the "prospects for new and longer term treatments and interventions for the condition" (RTC, 2004: 4-5; Van Wagner, Mykitiuk and Nisker, 2008).

The PGD policy of the ITA in Victoria employs the criteria set out in s 8(3) of the *Infertility Treatment Act 1995* that a "genetic abnormality or disease might be transmitted to a person born", but it does not define these terms (ITA, 2006b). The policy "entrusts" this determination to "the specialist with qualifications in human genetics", explicitly placing the physician in the role of "gatekeeper" with respect to PGD (ITA, 2006b: [4.2]). The ITA's approach to the regulation of PGD includes using a schedule of "Approved Genetic Testing", which outlines "routine" uses that do not require notification of application (ITA, 2006a).⁸ The ITA also sets out uses that "require approval on a case by case basis", such as sex-linked conditions involving "inconclusive evidence about the transmission of that

8 List A of the schedule to the Act discusses the "Use of PGD where women have already been admitted for treatment and where the purposes of PGD is to detect chromosomal imbalances in the cases: recurrent implantation failure; recurrent miscarriage; advanced maternal age; previous history of fetal aneuploidy; known carriers of chromosomal rearrangements". List B covers "Current use of PGD in Victoria where further notification to the Infertility Treatment Authority is not required": known carriers of chromosomal anomalies; determination of embryonic sex in certain specific conditions; and particular heritable single gene disorders.

condition", including Autism and Asperger's Syndrome (ITA, 2006a: List C). Conditions not listed in the schedule would require "prospective notification" to the authority (ITA, 2006a: 5). Approval for PGD in these circumstances requires fulfilment of the s 8(3) criteria as determined by a doctor specialising in genetics (ITA, 2006b: [6a] [6c]).

In Canada, access to PGD "is currently controlled by the medical profession" (CBS, 2005a: 2); however, "there are no Canadian standards or professional guidelines relating to the use of PGD in Canada" (HCARO, 2005: 6). Without formal regulation, decisions concerning PGD are made privately by the woman, or couple, and the doctor (CBS, 2005a: 2). Health Canada's report on the regulation of PGD distinguishes use for "medical/health reasons" from use for "non-health related traits, such as hair or eye colour". They use the "serious condition" standard as a generally accepted limitation to PGD, but acknowledge that seriousness would be "difficult to define" and that "there are many complex factors that need to be accounted for in this definition" (HCARO, 2005: 11; Van Wagner, Mykitiuk and Nisker, 2008).

New Zealand's PGD guidelines state that in order to perform PGD for "familial single gene", "familial sex-linked" (NECAHR, 2005: 5 s 2.4) and "familial chromosomal" disorders (NECAHR, 2005: 5), there should be "evidence that the future individual may be seriously impaired as a result of the disorder" (NECAHR, 2005: ss 1.3, 2.4, 3.2). Seriousness is not defined, but the guidelines provide that "[i]t is the responsibility of PGD providers, in collaboration with a clinical geneticist, to determine whether a disorder is likely to be serious in the offspring" (NECAHR, 2005). In this situation, the woman and her family do not participate in the determination of "seriousness" (Van Wagner, Mykitiuk and Nisker, 2008).

One can critique the "seriousness" standard from a number of perspectives:

Although PGD can confirm the presence of a genetic anomaly, it cannot predict the extent to which the *in vitro* embryo, if transferred into the womb and born alive, would be affected as a child or adult. (CBS, 2005a: 2)

This uncertainty becomes particularly problematic when PGD is used to diagnose "later onset" disorders such as Alzheimer's or "low penetrant" conditions (HCARO, 2005: 11; HGRP, 2006: 50). Further, understandings of what constitutes "suffering", what one might consider "detrimental effects", or a "life limiting" or "threatening" condition, are subjective and dependent on numerous factors relating to both the individuals and family involved, along with the particular condition at issue (HGRP, 2006: 37). The HFEA itself suggests that "these factors may be difficult to predict before the affected person is born" (2005: 11; Van Wagner, Mykitiuk and Nisker, 2008).

Evidence suggests that:

People with genetic disorders, their families and professionals have different views about which conditions give rise to a poor quality of life. In general, those who have a direct experience of living with a genetic disorder are likely to rate the quality of their lives more highly than would medically qualified professionals. (HGRP, 2006: 37)

The approach of the HFEA and Western Australia of considering the perceptions of the people seeking IVF and PGD in defining "seriousness", illustrates the subjectivity of such a determination (Van Wagner, Mykitiuk and Nisker, 2008). While the STC legislative review dismissed the use of the word "eugenics" by its critics "as an emotive term of abuse to obscure rational debate" (STC, 2005: 55), concerns about who defines what a "serious" condition is, and on what basis, have not been adequately addressed by either law or policy in any of the jurisdictions examined. Rather than confronting the complexity of the shifting nature of determining "seriousness" and its consequences, we contend that current legal and policy approaches ignore subjective considerations in favour of "medical" or "scientific" criteria. This focus seems to imply that "seriousness" can be defined outside the context of the lives and experiences of those undergoing ART and PGD (Van Wagner, Mykitiuk and Nisker, 2008).

The HGRP points out that in New Zealand, legislation and policy decisions have given professionals involved in PGD "a broad mandate to determine what constitutes a disorder that could cause serious impairment in a future child, and the likelihood of it happening" (2006: 236). The HGRP distinguishes

between the role of clinicians in determining the "likelihood of a disorder manifesting in prospective offspring", which they view as "generally unproblematic", and their role in stating "what constitutes a serious disorder" (HGRP, 2006: 236). This latter determination involves both "objective" considerations, including the age at which a disease emerges and the possibility of prevention and/or therapy, and "subjective" considerations, including the "experience of the prospective parents in relation to the condition" (HGRP, 2006: 236). Their report discusses the possibility that "by leaving such decisions in the hands of treating clinicians, rather than in those seeking the procedure, PGD cannot be represented as providing greater autonomy and reproductive freedom" (HGRP, 2006: 236).

Basing decisions about PGD on a "discussion between the people seeking treatment and the clinical team" is presented as a way to balance "respecting the views of those seeking PGD" and "preventing the use of technology for purposes that are widely considered to be unacceptable" (HFEA, 2005: 12). While we support the primacy of a woman's role in determining what reproductive choices are best for her and her family, we caution that respect for reproductive autonomy should not be invoked to allow policy-makers and clinicians to avoid complex and difficult questions about the potential implications of reproductive and genetic technologies on conceptions of "health" and "normalcy" (Van Wagner, Mykitiuk and Nisker, 2008). Questions about how the use of technologies such as PGD may affect social norms and ideas about family and being human (HGC, 2004: 20) or about how "reproductive choices are being made against a background of inadequate social support for, and widespread discrimination against, disabled people and people with genetic disorders" (HGC, 2004: 22 5.8) should not be obscured or limited to private discussions in the realm of the clinic (Van Wagner, Mykitiuk and Nisker, 2008).

What happens to the affected embryo?

Upon being identified as "affected", the post-PGD embryo is constructed to be incompatible with reproduction. Characterised as "unsuitable" (Australian Government, 2005: 169; STC, 2005: 23) or "unfit" (Australian Government, 2005: 175), the

affected embryo is generally assumed to be "discarded" (Australian Government, 2005: xvi; CBS, 2005a: 5, 7, 22; HFEA and ACGT, 2000: 8; HGRP, 2006: 7, 55, 320; NECAHR, 2004: 7), "allowed" (HFEA, 2005: 7) or "left" (CBS, 2005a) to perish, or that PGD will result in their "disposal" (HFEA and ACGT, 2000: 8) or "destruction" (HGC, 2004: 45; HGRP, 2006: 163). As the recent legislative review in Australia found:

Under current arrangements, embryos that are not suitable for implantation for any reason, including embryos that are found to have a genetic disease using preimplantation genetic diagnosis, are allowed to die and are not available for research. (Australian Government, 2005: 168)

However, recent debates in Australia suggest that the designation of "affected" embryo, may signal a shift in an embryo's purpose and/or value (Van Wagner, Mykitiuk and Nisker, 2008). On one hand, the value of an affected embryo for reproductive purposes is diminished, as it is assumed that the intention behind undergoing the diagnostic procedure is the avoidance of the transfer of an embryo affected by the genetic condition being tested for (Australian Government, 2005: 120; ITA, 2006b: [3]). On the other hand, however, the affected embryo becomes valuable in the context of research and training (Australian Government, 2005: xvi). Indeed, a recent ACART consultation on embryo research in New Zealand specifically discussed post-PGD embryos as one source of surplus embryos for research purposes (ACART, 2006: 13).

Just as for non-PGD embryos (Nisker and White, 2005), our research suggests that attempts to characterise post-PGD embryos may be driven by both the increasing demand for embryos for research purposes and ethical concerns about the use of human embryos (see discussions in Australian Government, 2005; CBS, 2005a; Van Wagner, Mykitiuk and Nisker, 2008). ACART proposes using post-PGD embryos for research, as "they may never be transferred to a woman's uterus" (ACART, 2006: 13). The presumption that post-PGD affected embryos would otherwise be "discarded" (Australian Government, 2005: xvi) featured prominently in the 2005 Australian legislative review. The RIHE Act has since undergone major amendments with respect to the availability of affected embryos

for research pursuant to the *Prohibition of Human Cloning for Reproduction and the Regulation of Human Embryo Research Amendment Act 2006* (Cth) (Patterson Act). A number of submissions made to the legislative review committee pointed to the lack of clarity in the RIHE Act on the status of post-PGD embryos deemed "unsuitable for implantation" (Australian Government, 2005: xvi, 31, 38, 76, 169).⁹ Several parties¹⁰ argued that post-PGD affected embryos should not be considered simply as "excess" embryos subject to the same consent and donation process outlined in the "ART Guidelines" that applies to embryos created by IVF, but which the couple no longer needs for reproduction (see NHMRC, 2004: [17.17]). Rather, parties argued that through policy and legal reforms to avoid their characterisation as "excess", such post-PGD affected embryos could be made available as "fresh" embryos for research and training (Australian Government, 2005: 120).

Submissions to the Review Committee consistently suggested that "fresh embryos" were "required" (Australian Government, 2005: 37) and that "abnormally fertilised" and "unsuitable" embryos "should be made available for research and training" (Van Wagner, Mykitiuk and Nisker, 2008). The Committee recommended that post-PGD embryos "diagnosed ... as being unsuitable for implantation should be permitted to be used under licence for research, training and improvements in clinical practice" (Australian Government, 2005: xvii). This recommendation was specifically aimed at addressing the unavailability of "fresh" (Australian Government, 2005: 38) embryos resulting from the 14-day "cooling-off" (Australian Government, 2005: 37) period applied to donations of "excess" embryos following IVF (NECAHR, 2004: s 17.7), which would be frozen and stored for 14 days before becoming available for research. The Committee relied on the assumption that affected embryos would "normally

9 The review notes that the lack of clarity does not necessarily exist in the States that have independent legislation in place to govern ART and PGD (2005, 169).

10 For the most part, parties made confidential submissions to the committee. Submissions made openly available include those made by: The Plunkett Centre for Ethics, Submission LRC550; Professor Martin Pera et al, Monash University, Submission LRC509 and Sydney IVF, Submission LRC 819. All are available online at <www.lockhartreview.com.au/submissions.html> (last accessed 10 May 2007).

be discarded" (Australian Government, 2005: 120), as they were deemed unsuitable for reproductive purposes, and therefore, that they need not be subject to "proper consent" procedures.

Using post-PGD embryos as a source of "fresh" embryos for research raises concerns that determinations made by those practicing PGD will be influenced by the demand for research embryos. Increased comfort with production of embryos through IVF for research purposes may serve to justify the expansion of PGD's application and to narrow the definition of a "suitable" or "unaffected" embryo in the interests of ensuring "fresh" embryos are available. In turn, our understanding of what kinds of conditions are compatible, or incompatible, with reproduction and health may be further shifted to exclude particular genetic characteristics (Van Wagner, Mykitiuk and Nisker, 2008).

Throughout the documents we surveyed, the presumption of selection against embryos affected by genetic conditions is widespread, despite ongoing debate about the implications of using PGD to make such determinations and the lack of transparency in deciding to which conditions PGD should be applied. Distinctions are being made between embryos to be used for reproduction and affected embryos based on problematic assumptions about the use and purposes of reproductive technologies (Van Wagner, Mykitiuk and Nisker, 2008). The assumption that "reproductive use" inherently means selecting an embryo free of a particular genetic condition or abnormality (Australian Government, 2005: 120) subtly shifts the meaning of reproductive use to exclude the conditions for which PGD is licensed and applied. However, it is the process of PGD itself, and the characterisations and determinations of clinicians about the presence of particular genetic markers or abnormalities in the post-PGD embryo, which construct the resulting lack of suitability in the embryo. In doing so, PGD and those applying it redefine reproduction to exclude the affected embryo (Van Wagner, Mykitiuk and Nisker, 2008).

Legislating suitability: the unsuitable embryo in law

The Patterson Act amends the RIHE Act, authorising modifications to the "proper consent" requirements for licenses to use

"unsuitable" embryos. The Act now defines "unsuitable for implantation, in relation to a human embryo" in s 7(1) in the following manner:

- (a) is diagnosed by preimplantation genetic diagnosis as unsuitable for implantation, in accordance with the *Ethical Guidelines on the Use of Assisted Reproductive Technology in Clinical Practice and Research* (2004), issued by the CEO of the NHMRC; or
- (b) is determined to be unsuitable for implantation in the body of a woman, in accordance with objective criteria specified in guidelines issued by the CEO of the NHMRC under the *NHMRC Act 1992* and prescribed by the regulations for the purposes of this paragraph.

The amendment directly responds to the Review's recommendations that ART embryos be deemed unsuitable according to "objective criteria", and through the use of PGD to detect "serious genetic defects" (Australian Government, 2005: xvi).¹¹ The Lockhart Review distinguishes objective determinations of PGD from the subjective nature of determining "when the embryo appears less healthy" (Australian Government, 2005: 17). The RIHE Act now legally sanctions PGD as an objective means of distinguishing the suitable embryo from the unsuitable (Van Wagner, Mykitiuk and Niskier, 2008).

Both the Lockhart recommendations, and now the amended law, imply that "health" is understood as the absence of the genetic conditions identified by PGD (Van Wagner, Mykitiuk and Niskier, 2008). This assumption about "health" allows the Review to avoid debate on the limitations of the genetic determinations of health (NECAHR, 2004: 14), as well as on the implications of PGD for people living with disabilities (NECAHR, 2004: 40, 61). Framing PGD as an "objective" way to determine "suitability", the Review fails to acknowledge and deal with the subjective factors that influence clinical decisions

11 The NHMRC has issued guidelines on the "objective criteria" on which decisions about suitability for implantation are to be made based on morphologic characteristics. They can be found online at: <www.nhmrc.gov.au/embryos/stemcells/_files/objective_criteria.pdf> (last accessed 23 June 2008). "Contextual Information" to these guidelines is available online at: <www.nhmrc.gov.au/embryos/stemcells/_files/contextual_info.pdf> (last accessed 23 June 2008).

about when PGD should be applied, and for what purposes (Van Wagner, Mykitiuk and Nisker, 2008). The Review relies on the assumption that affected embryos would "never" (Australian Government, 2005: 120) be used for reproductive purposes to justify their availability to researchers.

We suggest that, in light of the problematic nature of defining and describing "seriousness", "health" and "quality of life" with respect to genetic conditions, it is the application of PGD itself which produces the inevitability that embryos with particular genetic conditions will never be transferred to the woman, not the inherent or biomedical incompatibility of particular genetic characteristics with reproduction (Van Wagner, Mykitiuk and Nisker, 2008). The new law of Australia makes the practice of ART professionals in administering PGD the source of an "objective" (Australian Government, 2005: 169) assessment of suitability. Such determinations become the means through which "fresh" embryos are made available for "research, training and improvements in clinical practice" (Australian Government, 2005: 169; Van Wagner, Mykitiuk and Nisker, 2008).

Section 24 of the RIHE Act sets out the licensing requirements for researchers to use excess ART embryos. The amended sub-s (8) states that a licence may provide for a modified application of the proper consent guidelines to the use of "unsuitable" embryos (s 8(a)). The amendment does not specifically remove the cooling-off period for post-PGD affected embryos, as recommended by the Lockhart Review; however, it includes the following note: "[f]or example, the guidelines could apply to a particular licence in a modified form, to alter the cooling-off period required in relation to the use of excess ART embryos that are unsuitable for implantation" (Patterson Act s 24(8)).

Conclusion

While much of the focus in debates about research on *in vitro* embryos has centred on whether an embryo's moral status should be affected by the processes through which an embryo has been created and the intention behind that creation (CBS, 2005a: 36), our analysis suggests another source of distinction is emerging through the use of PGD – one which determines moral

and ethical status on the basis of the embryo's genetic classification as "affected" or "unaffected" by genetic conditions (Van Wagner, Mykitiuk and Nisker, 2008). Driven by the need for sources of embryos, particularly "fresh" embryos, for research and training activities, assumptions about the objectivity and the purposes of PGD are allowing clinicians, researchers and policy-makers to circumvent debates about the social dimensions of (re)defining reproduction to exclude a growing number of genetic characteristics and conditions (Van Wagner, Mykitiuk and Nisker, 2008). The consequences of this conceptual shift could be profound for people living with disabilities, their families and broader society. Scholars and policy-makers must expose assumptions behind notions of health, normalcy and reproductive choice, and must examine them to ensure that new practices and technologies benefit all women and their families, not just those who fit within the status quo.